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Hyalo-retinopathy in the clefting syndrome

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The hereditary association of cleft palate, hypoplastic maxilla, vitreo-retinal degeneration, and myopia, is known as clefting syndrome, and was first reported over 30 years ago.

Knobloch and Layer (1972) presented a series of pedigrees with clefting syndrome and associated skeletal abnormalities. The purpose of this report is to present a further pedigree with clefting syndrome, to stress the seriousness of the ocular condition and the problems of management.

Material

Four patients of the same family were managed at the Retinal Unit of the High Holborn branch of Moorfields Eye Hospital over a period of 10 years (1963 to 1973). The family consisted of a father, aged 37, and three sons (only children), aged 14, 13, and 10 years. As far as is known the father's parents and siblings had normal eyes. They were not examined as they are scattered around the world.

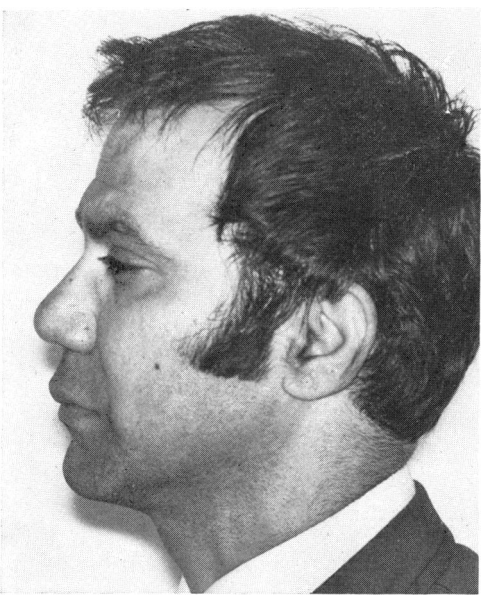

FIG. A *Appearance of Case* 1

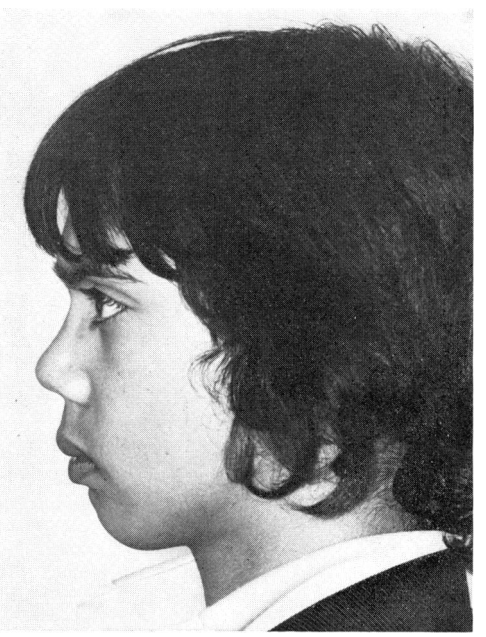

FIG. B *Appearance of Case* 2

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CLINICAL FEATURES

The physiognomy of all four cases was similar (Figs A to D). The strong family resemblance was characterized by maxillary hypoplasia, and a broad face with depressed nasal bridge; the father had a normal nasal structure. Details of the palatal and skeletal abnormalities occurring in this pedigree are shown in Table I (overleaf).

OPHTHALMIC FINDINGS

The three oldest members of the family have had bilateral retinal detachments which occurred in the first and second decade. At least three, and probably four, of the six eyes developed giant tears of approximately 180° in circumference.

The most striking feature was the vitreous degeneration which is characteristic of clefting syndrome. The pathological changes ranged from early vitreous syneresis to marked degeneration with membrane formation. In Case 1 the vitreal changes were minimal, in Case 2 marked, and in Case 3 moderate. In Case 4 no vitreal abnormality was detected when the patient was first examined at the age of 6 years, but during the last 2 years vitreous strands have developed.

The retinal changes ranged from normal to microcystoid with retinoschisis. All four patients were myopic (range $-1{\cdot}5$ to -15 D). In only one case (Case 2) was glaucoma present. This was associated with an angle anomaly.

The father is rapidly developing a cortical cataract in his only eye. This agrees with the findings of Frandsen (1966), who noted an early onset of lens opacities in these patients.

Surgical procedures (Table II, pp. 100 and 101)

Case 1

This patient had already had bilateral retinal detachment surgery before he was seen at Moorfields Eye Hospital. The right eye developed a detachment when he was 12 years old and had to be enucleated after the development of a postoperative endophthalmitis. The retina in the left eye detached 5 years later and was successfully treated by release of subretinal fluid and diathermy.

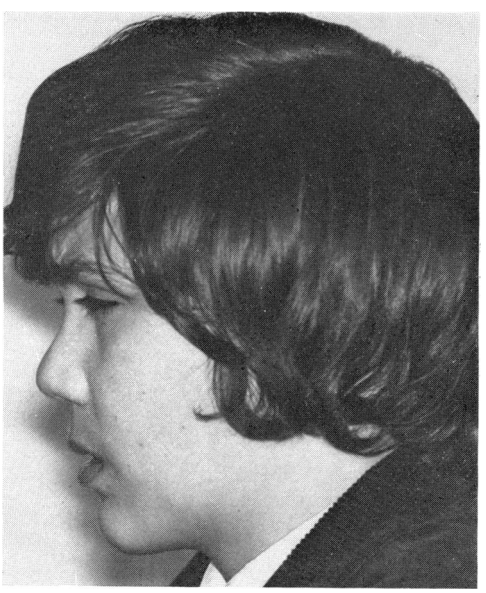

FIG. C *Appearance of Case 3*

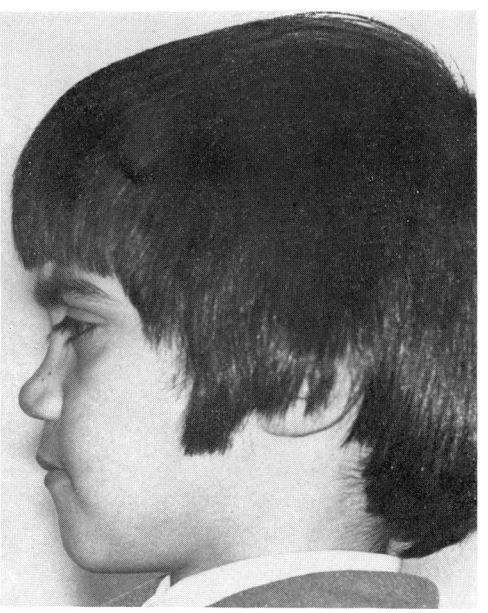

FIG. D *Appearance of Case 4*

Table I *Palatal and skeletal abnormalities*

Case no.	1	2	3	4
Age (yrs)	37	14	13	10
Maxilla	Broad	Hypoplastic	Hypoplastic	Hypoplastic
Jaw	Normal	Normal	Normal	Normal
Epicanthus	Nil	Very slight	Slight	Nil
Interpupillary distance (cm.)	7	6·5	6·5	6
Nose	Normal	Depressed bridge	Depressed bridge broad nose	Depressed bridge broad nose
Palate	Complete cleft (repaired)	Complete cleft (repaired)	Complete cleft (repaired)	Complete cleft (repaired)
Upper lip	Normal	Normal	Normal	Normal
Hands	Proximal interphalangeal joints enlarged	Proximal interphalangeal joints slightly enlarged	Interphalangeal joints normal, tapering fingers, hyperextensible	Interphalangeal joints normal, long tapering fingers, hyperextensible
Feet	Talipes equinovarus Flat arch (mild)	3rd toe curled in Arch normal	Slight pes cavus	Long 2nd and 3rd toes, 4th toe curled in
Elbows	Hyperextensible	Hyperextensible	Hyperextensible	Hyperextensible
Knees	Hyperextensible	Hyperextensible	Hyperextensible	Hyperextensible

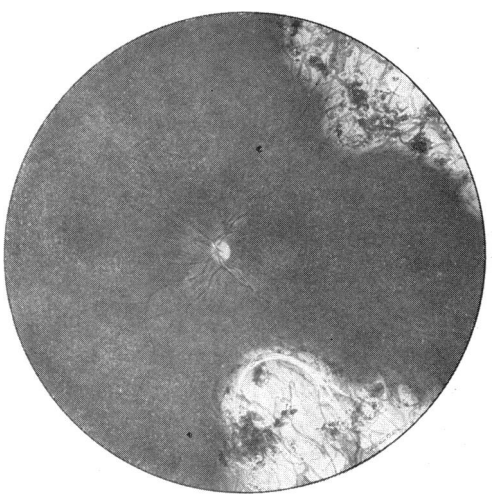

FIG. 1 *Left eye of Case* 1, *showing flat retina after surgery in* 1952

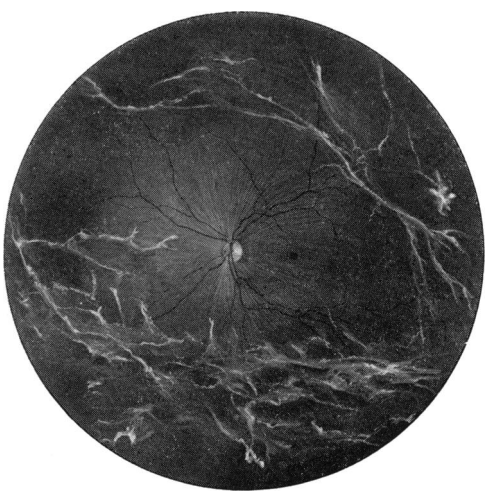

FIG. 2 *Left eye of Case* 2, *showing marked vitreoretinopathy*

FIG. 3 *Left eye of Case 2, showing total retinal detachment with a giant tear*

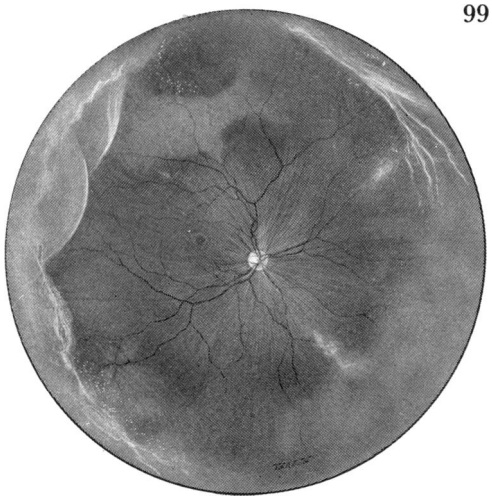

FIG. 4 *Right eye of Case 3, showing moderate vitreo-retinopathy with retinoschisis*

FIG. 5 *Right eye of Case 3, showing total detachment with a giant tear*

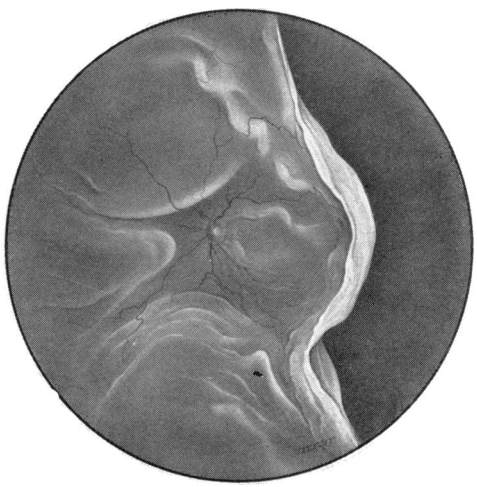

FIG. 6 *Left eye of Case 3, showing total retinal detachment with a giant tear*

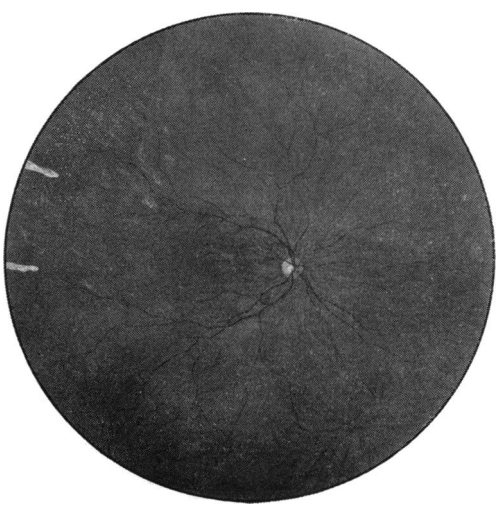

FIG. 7 *Right eye of Case 4, showing early vitreo-retinopathy*

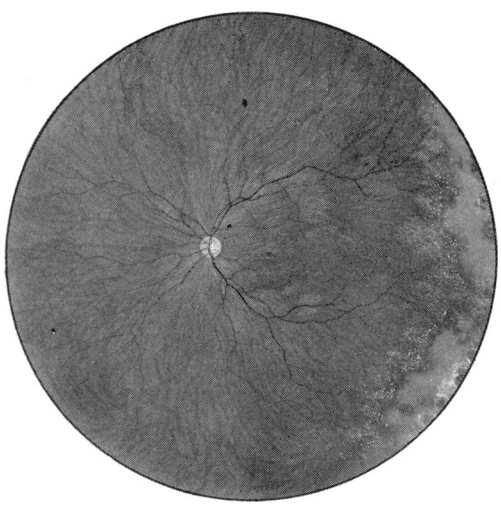

FIG. 8 *Left eye of Case 4, showing early vitreo-retinopathy*

The three eyes with giant retinal tears were operated on by members of the retinal unit at Moorfields Eye Hospital.

Case 2

This patient presented at the age of 6 years with a mature cataract in the right eye, and no perception of light. The history suggests that a retinal detachment developed in this eye approximately 18 months before presentation, and that a complicated cataract developed subsequently. No view of the fundus was possible at the time of presentation.

The left retina became detached when he was 12 years old. This was successfully treated by encirclement with a silicone strap, cryotherapy, and retinal incarceration by the suction technique as described by Howard and Gaasterland (1970).

Case 3

In this patient the retina of the left eye became detached at the age of 7 years. It was unsuccessfully

Table II *Ocular findings and treatment*

Case no.	1		2	
Age (yrs)	37		14	
Year of presentation	1960		1963	
Eye	Right	Left	Right	Left
Refractive error	–	−15·00	–	−8·00
Visual acuity (on presentation)	Anophthalmos	6/12	No perception of light	6/9
Pre-detachment findings		?	? (mature cataract)	
Figure no.		1		2, 3
Operations	*1948* Eye enucleated at age 12 years after retinal detachment surgery	*1952* Surface diathermy	Nil	*1971* Encirclement Cryotherapy Retinal incarceration
Present visual acuity		6/12 Lens opacities	No perception of light Mature complicated cataract	6/24
Present state of retina		Flat	Probably total retinal detachment	Flat

treated by encirclement with a silicone strap, plombage, and cryotherapy. Massive vitreous retraction resulted and no further surgery was attempted.

The right eye developed a retinal detachment at the age of 12 years. It was treated by encirclement with a silicone strap, cryotherapy, and retinal incarceration in the first instance. This was unsuccessful, and a further attempt at incarceration was made which also failed. Massive vitreous retraction resulted and intravitreal liquid silicone oil was injected 5 months later. Unfortunately, no functional improvement has been achieved.

Case 4

The left eye has recently been encircled prophylactically, and the right eye has been treated by 360° cryotherapy.

Discussion

This family shows the typical features of a clefting syndrome.

Of the eight eyes affected only two remain undetached. These belong to the youngest member of the family, who is only 10 years old at the time of writing, but who already has

3		4	
13		10	
1965		1969	
Right	Left	Right	Left
−1·50	−1·50	−4·50	−4·50
6/18	Perception of light	6/12	6/12
4, 5	6	7	8
1972 (1) Encirclement Cryotherapy Retinal incarceration (2) Retinal incarceration (repeated) (3) Intravitreal silicone injection	*1967* Encirclement Cryotherapy Plombage	360° cryotherapy	*1973* Prophylactic encirclement
Hand movements	No perception of light	6/12	6/12
Total retinal detachment	Total retinal detachment	Flat	Flat

marked vitreo-retinopathy in both eyes. In view of the course of events in the other members of the family, prophylaxis has been performed on both eyes.

It is interesting that the retinal detachments developed at an uncommonly early age, with a very high incidence of giant retinal tears extending for 180°. The progressive nature of the vitreal degenerative changes was also a striking feature in these patients.

As in the majority of cases with such extensive tears, the surgical results were very disappointing, although one patient had a satisfactory result. We would, therefore, like to stress the poor prognosis of this syndrome, and urge frequent ocular examination in these cases, in order that early prophylaxis may be carried out.

Summary

Four cases of clefting syndrome are described in a man and his three sons. The serious nature of the vitreo-retinal changes and the poor surgical results of retinal detachment surgery are emphasized. A plea is made for early prophylaxis.

We are greatly indebted to Mr. J. R. Hudson for allowing us to describe cases under his care.

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